

Diagnosis and treatment of paroxysmal kinesigenic dyskinesia in a 15-year-old boy

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EDITOR'S KEY POINTS

- Paroxysmal kinesigenic dyskinesia (PKD) is a rare condition with an estimate prevalence of 1 in 150 000 and an average time to diagnosis of almost 5 years. Although PKD is a rare condition, it is important that primary health care providers are aware of its clinical presentation, as PKD can be easily dismissed as a psychosomatic or psychiatric illness.
- Paroxysmal kinesigenic dyskinesia is characterized by abnormal involuntary movements precipitated by a sudden movement or startle.
- It is important to recognize PKD, as it typically responds well to anticonvulsant medications. Carbamazepine is considered the first-choice treatment, as phenytoin has a less favourable drug profile.

POINTS DE REPÈRE DU RÉDACTEUR

- La dyskinésie kinésigénique paroxystique (DKP) est un problème rare dont la prévalence estimée est de 1 sur 150 000 et il faut en moyenne près de 5 ans pour la diagnostiquer. Même si la DKP est un problème rare, il importe que les médecins de soins primaires soient au courant de sa présentation clinique, parce que la DKP peut facilement être prise à tort pour une maladie psychosomatique ou psychiatrique.
- La dyskinésie kinésigénique paroxystique se caractérise par des mouvements involontaires anormaux déclenchés par un mouvement soudain ou un sursaut.
- Il est important de reconnaître la DKP car elle répond habituellement bien aux médicaments anticonvulsifs. La carbamazépine est considérée comme le traitement de première intention, parce que la phénytoïne a un profil pharmacologique moins favorable.

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Case

A 15-year-old, right-handed boy presented with a 1-year history of involuntary movements primarily involving the right side of his body. These movements would last approximately 20 to 30 seconds and occurred 10 to 15 times a day. He described being able to sense when they would occur. He never lost consciousness during these spells, but he was unable to speak. The movements were triggered by voluntary activities like getting out of bed or a chair after a period of physical rest. He was able to voluntarily induce an episode.

The patient has always been in good health and did not take any medications. His younger brother was diagnosed with rolandic epilepsy following 1 seizure. There was no other family history of involuntary movements or seizures.

He was an excellent grade 10 student. He had become less active in sports because his movement disorder was interfering with physical activity.

Results of physical and neurologic examinations were normal. Two episodes of these involuntary movements were triggered in the office when he jumped off the examination table. He displayed involuntary dancelike and writhing movements involving the arms, legs, and face, and they were more noticeable on the right side. He was fully aware of his environment and was surprisingly able to avoid physical injury to himself.

After witnessing the episodes, the patient was referred to a neurologist as well as a psychiatrist.

Normal results were reported for complete blood count; random glucose, alanine transaminase, thyroid stimulating hormone, creatinine, electrolyte, creatine phosphokinase, total protein, and calcium levels; electroencephalography; and unenhanced magnetic resonance imaging.

The neurologist also witnessed an episode and diagnosed paroxysmal choreoathetosis. The patient was started on 100 mg of controlled-release carbamazepine twice daily. The patient was advised to increase the dose if symptoms were not improving and to contact the office if his progress was not satisfactory.

The patient indicated that his involuntary movements completely resolved immediately after starting the medication, although he would occasionally have a feeling of apprehension that another spell would occur.

The appointment with the psychiatrist was cancelled, as his symptoms resolved quickly with a low dose of controlled-release carbamazepine.

Discussion

Paroxysmal kinesigenic dyskinesia (PKD) is a rare condition characterized by abnormal involuntary movements that are precipitated by a sudden movement or startle.¹ While our patient presented with the choreoathetosis type of movement, a case series of 121 affected individuals¹ and a report of 26 patients² with PKD suggest that most patients present primarily with dystonic movements. A few patients might present with ballismus, hyperkinesias, and combinations of abnormal movements.¹ Speech involvement might be seen with paroxysmal nonkinesigenic dyskinesia (PNKD) and can be used as a distinguishing feature to help differentiate between PKD and PNKD³; however, disturbances such as dysarthria and anarthria as a result of facial muscle involvement were found in 8 of 26 (31%) patients with PKD in one case series.²

The prevalence of PKD is unknown, as epidemiologic data are not available owing to the rarity of paroxysmal dyskinesias,⁴ and many cases are not recognized. One author estimates a prevalence of 1 in 150 000.³ Lack of recognition results in delayed diagnosis and the average time to obtain a correct diagnosis in one study of 121 individuals was 4.8 years.¹ Hence, it is important for physicians to be aware of this condition, as the initial presentation can be easily missed if not correctly recognized.

Paroxysmal dyskinesias are classified according to their triggers, the duration and frequency of attacks, the effectiveness of medication, and associated syndromes.³ Paroxysmal kinesigenic dyskinesia is the most frequently encountered subtype of paroxysmal dyskinesia. The other syndromes are PNKD,³ paroxysmal exertion-induced dyskinesia,³ paroxysmal hypnogenic dyskinesia (frontal lobe epilepsy),³ and paroxysmal dystonia or torticollis in infancy.⁵

The assessment of 121 patients with PKD¹ found that episodes typically lasted less than 1 minute, and a “premonitory sensation of imminent attack” was reported by 82% of the patients. A frequency of more than 20 attacks per day was reported by 34% of the patients, while 47% reported a frequency of 1 to 20 per day. All patients reported sudden movements as the trigger. The most common type of movement observed in PKD was dystonia (57%), while chorea was reported by 6% of the patients and ballismus by 1%. A mixed picture was reported in 33% of the patients. Unilateral movements were observed in 36% of the patients, unilateral movements that alternated sides were observed in 12%, bilateral movements were observed in 35%, and either unilateral or bilateral movements (ie, an episode would only have either unilateral or bilateral movements) were observed in 18%.

Most cases are idiopathic, although familial cases occur and usually have an autosomal dominant inheritance pattern.^{3,4} In general there is an increased familial incidence of seizure disorders among patients with PKD.^{1,3}

Diagnosis

The diagnosis is based primarily on history and clinical observation, confirmed by normal imaging and laboratory test results. Clinical criteria based on the assessment of 121 individuals with PKD are outlined in **Box 1**.¹

Paroxysmal nonkinesigenic dyskinesia is the main differential diagnosis to consider. Paroxysmal nonkinesigenic dyskinesia can present with similar symptoms and signs; however, the attacks are not triggered by voluntary movement (kinesigenic).^{3,4} Also, the duration of the attacks is longer with PNKD, as an attack might last minutes to hours.^{3,4} Patients with PKD might have 1 to 20 attacks a day, while those with PNKD tend to have a lower frequency such as less than or up to 1 episode per week.³ Patients with PNKD are also less likely to respond to antiepileptic medications, although some might respond to clonazepam.⁴

Symptomatic paroxysmal dyskinesias do occur and usually are associated with other neurologic symptoms and abnormalities affecting older individuals.⁴

In many instances the patient presenting with dyskinesia might seem to have a condition with a psychogenic origin, which might be the case in PNKD but not in PKD.⁴ It is important to recognize the presenting features of PKD to facilitate a timely diagnosis and initiate appropriate treatment.

Conclusion

Although PKD is a rare condition, it is important that primary health care providers are aware of its clinical presentation, as PKD can be easily dismissed as a psychosomatic or psychiatric illness. Knowing the clinical presentation will help to assess the patient, determine suitable specialists to consult, and treat patients when appropriate.

Patients and caregivers should be informed about the benign course of the disease and the tendency to have decreased attack frequency with age. These patients typically respond well to anticonvulsant


Box 1. Clinical criteria for PKD

The clinical criteria for PKD are as follows:

- Identified kinesigenic trigger for the attacks
- Short duration of attacks (< 1 min)
- No loss of consciousness and no pain during attacks
- Exclusion of other organic diseases and normal neurologic examination results
- Control of attacks with an antiepileptic drug such as carbamazepine or phenytoin, if tried
- Age at onset between 1 and 20 y if no family history of PKD. Age criteria might not be as important in the familial cases. Cases with older age of onset have been described in instances where there is a family history of PKD

PKD—paroxysmal kinesigenic dyskinesia.

Data from Bruno et al.¹

medications.^{1,4} A literature review⁴ suggests a daily dose of 200 to 400 mg of extended-release carbamazepine for adults and 1.5 to 2 mg/kg for children. Carbamazepine is considered the first-choice treatment, as phenytoin has a less favourable drug profile. Other medications described in case reports and case series as alternative treatments include lamotrigine, levetiracetam, oxcarbazepine, topiramate, valproate, and benzodiazepines such as clonazepam.^{1,4} 

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Competing interests

None declared

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